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Retinoblastoma

From Wikipedia, the free encyclopedia.

Retinoblastoma is a cancer of the retina. It is caused by a mutation in the Rb-1 protein. It occurs mostly in younger children and accounts for about 3% of the cancers occurring in children younger than 15 years. The estimated annual incidence is approximately 4 per million children [1]

(http://www.cancer.org/docroot/CRI/content/CRI_2_4_1X_What_are_the_key_statistics_for_retinoblastoma_37.asp?sitearea=).

The tumor may begin in one or both eyes. Retinoblastoma is usually confined to the eye but can spread to the brain via the optic nerve.

Retinoblastoma may be hereditary (genetically inherited) or nonhereditary. The hereditary form may be in one or both eyes, and generally affects younger children. Retinoblastoma occurring in only one eye is often not hereditary and is more prevalent in older children. When the disease occurs in both eyes, it is always hereditary. Because of the hereditary factor, patients and their brothers and sisters should have periodic examinations, including genetic counseling, to determine their risk for developing the disease.

A statistical study by Dr Alfred G. Knudson in 1971 led to a hypothesis (later known as the Knudson hypothesis) about why some retinoblastomas are hereditary and others occur by chance. This hypothesis led to the first identification of a tumor suppressor gene by a team led by Dr Thaddeus P. Dryja in 1986. Knudson won the 1998 Albert Lasker Medical Research Award for this work.

Hereditary retinoblastoma is caused by an inherited mutation in a single copy of the Rb1 gene. The remaining functional copy prevents most retinal cells from becoming cancerous. However, one or more cells in the retina are likely to undergo a spontaneous loss of this functional copy, causing those cells to transform into cancer. This loss of the second copy of Rb1 is termed loss of heterozygosity, a frequent event in cancer for which retinoblastoma is the canonical example.

The patient's choice of treatment depends on the extent of the disease within and beyond the eye. Smaller tumors can be removed with laser surgery, thermo-, or cryotherapy.

Genetic testing can identify the mutation that lead to the development of retinoblastoma. Testing in unilateral cases can identify the 15% of unilateral cases with a germline mutation, indicating risk in future children. Testing amniotic cells in an at-risk pregnancy can identify a fetus with the mutation, which can then be delivered early before retinal cells have fully developed and before tumors arise. This early treatment can lead to a fully sighted bilaterally affected patient.

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See also

- Eye cancer

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- OMIM 180200 (<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=180200>)
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